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Case Report

Agenesis of the Corpus Callosum with Rare Dysmorphic Features

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Abstract: Agenesis of corpus callosum is a common congenital or developmental malformation of brain. This is a case report of 13 year old female presented with delayed milestones, mental retardation, refractory hypertension and pallor since childhood. She had dysmorphic facies in the form of thick eyebrows, hypertelorism, low anterior hairline, hirsuitism, broad nose, thickened lips and low set ears. Upper limbs examination revealed brachydactyly and prominent finger pads. Lower limbs showed hyperkeratotic plaques with rough skin broad feet and short 4^{th} & 5^{th} toes with generalized hypertrichosis. Dignosis reported association with rare dysmorphic features, hypertrichosis and refractory hypertension with normal karyotyping. It is important to be aware of the features of agenesis of the corpus callosum, to distinguish it from other syndromes and instigate a search for further, more serious, anomalies with which it may be associated.
Keywords: Agenesis, corpus callosum, hypertrichosis, refractory hypertension, dilated ventricules.

INTRODUCTION

Agenesis of corpus callosum is a common congenital or developmental malformation of brain [1]. Incidence varies from in the general population, its estimated prevalence is 3-7 per 1000 birth, while in children with developmental disabilities it is 2-3 per 100 [2, 3]. It is often associated with other malformations and genetic syndromes Organ systems other than CNS, particularly the musculoskeletal and genitourinary systems, may be affected [4-7]. Agenesis may be complete, partial or atypical. Though it may be seen in asymptomatic individuals, it's generally considered a potential marker for neurological impairment and prognosis is frequently related to other associated abnormalities. Here we present a case of partial agenesis of corpus callosum with dysmorphic features.

CASE REPORT

A 13 year old female, second born child of a non-consanguineous marriage, presented with delayed milestones, mental retardation, refractory hypertension and pallor since childhood. She was treated for urinary calculi and hydronephrosis at the age of 7 years. The family and perinatal history was essentially normal.

On examination child had dysmorphic facies in the form of thick eyebrows, hypertelorism, low anterior hairline, hirsuitism, broad nose, thickened lips and low set ears. Upper limbs examination revealed brachydactyly and prominent finger pads. Lower limbs showed hyperkeratotic plaques with rough skin broad feet and short 4^{th} & 5^{th} toes with generalized hypertrichosis. Tanners SMR staging revealed pre-pubertial status of the development.

Vitals-Pulse was 82/min, Blood pressure was 180/140mm Hg (Rt upper limb), respiratory rate of 32/min. Anthropometry - head circumference 52 cm, weight- 20 kg, arm span 123 cm, height – 122 cm with US: LS ratio- 0.96.

Investigations revealed hemoglobin-5.2g/dl. TLC-7500 cells/cumm, with Polymorphs -75%. lymphocytes- 22%, Eosinophils – 2%, monocytes 1%. Pheripheral smear showed microcytic hypochromic anemia with relative neutropenia. Urine examination showed albumin (+), urinary calcium 20mg/dl, specific gravity- 1010, blood urea -25mg/dl, serum creatinine -1mg/dl, uric acid -4.2mg/dl. Calcium–creatinie ratio, serum sodium, potassium, calcium, phosphorus, pre-albumin and thyroid function tests were within normal range.

USG and color Doppler showed right kidney measured 6.6 X 2.9 cm, slightly decreased in size when compared to left, with increased echogenicity and scarring. Left kidney measured 7.3X 3.9 cm with increased echogenicity of cortex. Bilateral renal arteries were not visualized, bilateral intra- renal arteries shows normal color flow. IVU showed right kidney smaller than left with normal excretion of contrast into excretory collecting system.

ECHO showed concentric LVH with dilated LV, moderate LV systolic dysfunction. Fundoscopy revealed grade III hypertensive retinopathy, MR1 brain showed widely separated lateral ventricles with partial agenesis of corpus callosum and mild dilatation of the supratentonal ventricular system. Karyotyping was 46 XX. Metabolic screening of urine for mucopolysaccharides and oligosaccrides were negative.

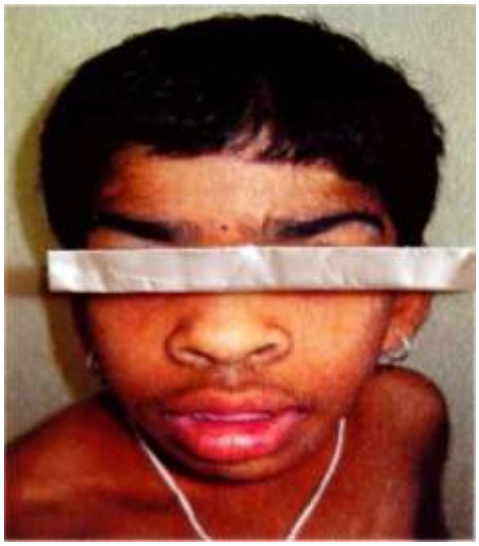

Fig. 1: Face-showing low anterior hairline, thick eyebrows, hypertelorism, hirsuitism, thickened lips, broad nose with low set ears

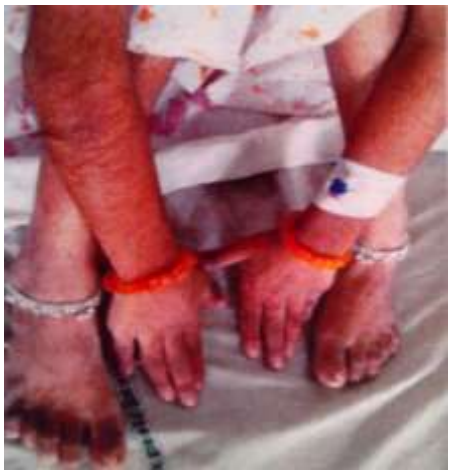

Fig. 2: Upper limbs showing brachydactyly & hypertricosis. Lower limbs with hyperkeratotic plaques with rough skin, broad feet and short 4th & 5th toes

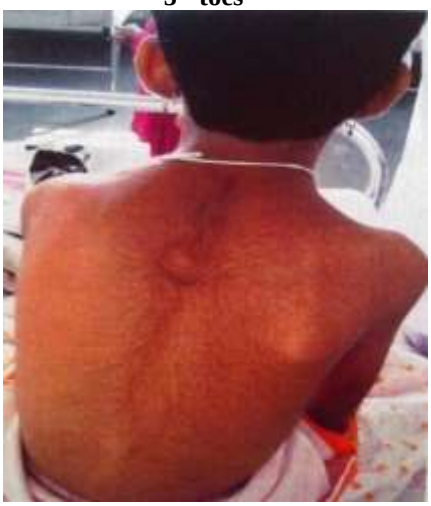

Fig. 3: Hypertrichosis

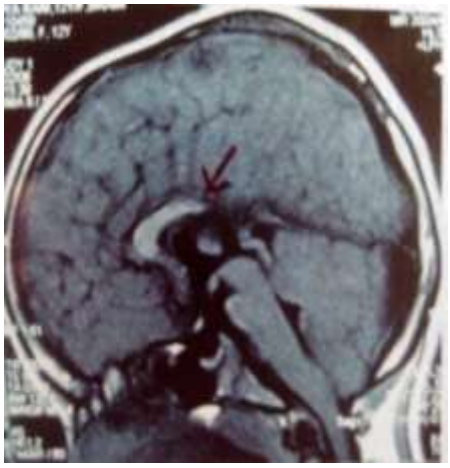

Fig. 4: Partial agenesis of the corpus callosum seen in Sagittal T1-weighted MRI of the brain. The genu and anterior body of the corpus callosum are visualized, whereas the posterior body, splenium, and the rostrum are absent.

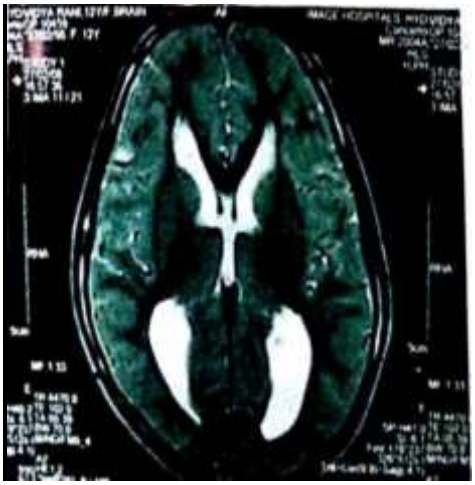

Fig. 5: Mildly dilated lateral ventricles seen in axial T 2-weighted MRI of the brain

Based on clinical findings such as developmental delay, mental retardation, dysmorphic facies and MRI showing partial agenesis of corpus callosum and mildly dilated ventricules with normal karyotyping -a diagnosis of Non -Syndromic partial agenesis of corpus callosum with dysmophic features was considered.

DISCUSSION

The corpus callosum develops from the lamina reunions in the telencephalon. It is the major axonal commissure of the brain connecting the two cerebral hemispheres and providing communication between the cortical and sub cortical neurons connecting one hemisphere with the corresponding part of the other hemisphere. It comprises of four parts: rostrum, genu, splenium and the body. It is essential for learning, discrimination, sensory experience, memory, synchronicity of sleep [8].

A wide range of developmental malformations can affect the corpus callosum, ranging in severity from total absence (agenesis) to lesser degrees of deficiency (hypoplasia) involving only the splenium. It may be complete, partial, or atypical. Embryonic development proceeds in an anterior to posterior direction, so if an injury occurs in a developing brain during the formation of the corpus callosum, the anterior part is usually present but the posterior part is deficient [8].

The clinical manifestations of callosal agenesis may be described under two headings: non-syndromic and syndromic. Non-syndromic forms are the most common. An unknown, though probably small proportion of patients are completely asymptomatic. Commonly, their condition is incidentally discovered during neuroimaging. Patients may present neurological problems, such as seizures (60%), intellectual impairment (70%), psychosis and cerebral palsy [9, 10]. However, these conditions are believed to be caused by abnormalities in associated cerebral anomalies rather than in the corpus callosum per se. Macrocephaly may occour as a result of hydrocephalus and is sometimes associated with interhemispheric cysts [11].

A number of syndromes may be associated with agenesis of corpus callosum. Some of the more common ones include Arnold-Chiari malformation, Dandy-Walker syndrome, Aicardi's syndrome, Andermann syndrome, Acrocallosal syndrome, schizencephaly, and holoprosencephaly and several of the trisomies [11].

So far many cases have been reported worldwide with partial or complete agenesis of corpus callosum associated with syndromic features, chromosomal abnormalities and inheritance pattern in form of autosomal dominant, autosomal recessive or X linked recessive pattern. Most of the cases have manifestations ranging from normal to severe mental retardation, seizures, hydrocephalus and psychomotor delay [12].

This case has been reported because of association with rare dysmorphic features, hypertrichosis and refractory hypertension with normal karyotyping. It is important to be aware of the features of agenesis of the corpus callosum, to distinguish it from other syndromes and instigate a search for further, more serious, anomalies with which it may be associated.

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